

Ordering Guidance: Genetic Testing for non-FH Hereditary Dyslipidemias

For further information on ordering genetic testing for Familial Hypercholesterolemia (FH), please refer to [Ontario Health's Ordering Guidance for Genetic Testing for FH](#)

Non-FH Dyslipidemia Testing Available in Ontario:

- Dyslipidemia multigene panels:
 - Hypertriglyceridemia, Low LDL-C, and Low HDL-C
- Disease Specific Test:
 - Cerebrotendinous xanthomatosis (single gene test)

Identifying Eligible Patients and Selecting a Genetic Test

Detailed eligibility criteria are outlined in the Hereditary Dyslipidemia Genetic Testing Recommendations ([Genetics Guidance](#)). Please see the table below for guidance on which test to consider:

Lipid Disorder	Eligibility Criteria	Recommended Genetic Test
Hypertriglyceridemia (HTG)*	<ul style="list-style-type: none"> • Fasting triglyceride levels ≥ 10 mmol/L on 3 separate occasions AND • Patient is under the age of 18 years 	Hypertriglyceridemia Panel
Low Low-Density Lipoprotein-C (Low LDL-C)	<ul style="list-style-type: none"> • Personal history of LDL-C levels ≤ 0.5 mmol/L at any age OR extremely low or undetectable apo B levels AND/OR • Clinical characteristics suggestive of a monogenic hypobetalipoproteinemia or abetalipoproteinemia 	Low LDL-C Panel
Low High-Density Lipoprotein-C (Low HDL-C)	<ul style="list-style-type: none"> • Personal history of HDL-C levels ≤ 0.5 mmol/L at any age AND/OR • Clinical characteristics suggestive of Tangier disease, LCAT deficiency, or apo A1 deficiency 	Low HDL-C Panel
Cerebrotendinous xanthomatosis (CTX)	<ul style="list-style-type: none"> • Elevated plasma cholestanol AND/OR • Clinical characteristics suggestive of CTX 	<i>CYP27A1</i> single gene test

* Severe hypertriglyceridemia is usually multifactorial in nature, with genetic testing not indicated in most cases. Please refer to the Hereditary Dyslipidemia Genetic Testing Recommendations document for further information on when to consider genetic testing for this condition.

Ordering Genetic Testing

Genetic testing can be arranged by a qualified geneticist or genetic counsellor. It is recommended that lipid specialists and/or endocrinologists who order genetic testing work in collaboration with a genetics clinic to establish appropriate testing and result disclosure pathways.

1. Complete a [requisition](#) from London Health Sciences Centre
2. Make a plan to review results; results will be sent to the ordering clinician in 8-10 weeks

Additional Support

For further assistance in test selection, test ordering, result interpretation, and management recommendations, please consider the following options as needed:

- Send a referral to your local genetics clinic ([Genetics Clinics in Ontario](#))
- Submit an eConsult request to connect with a genetics specialist through [OTNhub](#)